

CLAIMS

SUB A' 1. A method of diagnosing a mammal for an increased likelihood of developing a disease of the eye, said method comprising analyzing nucleic acid of said mammal to determine whether said nucleic acid contains a mutation in a FREAC3 gene, wherein the presence of said mutation is an indication that said mammal has an increased likelihood of developing a disease of the eye.

2. The method of claim 1, wherein said mutation is a missense mutation.

3. The method of claim 2, wherein said mutation results in a truncated protein.

4. The method of claim 1, wherein primers are used for detecting said mutation.

5. The method of claim 1, wherein said analyzing includes detecting the loss of a recognition site for a restriction endonuclease.

6. A kit for the analysis of FREAC3 nucleic acid, said kit comprising nucleic acid probes for analyzing the nucleic acid of a mammal, wherein said analyzing is sufficient to determine whether the mammal contains a mutation in said FREAC3 nucleic acid.

543 B'
7. A method of diagnosing a mammal for an increased likelihood of developing a disease of the eye, said method comprising detecting the presence of a mutant FREAC3 polypeptide in said mammal, wherein the presence of said mutant FREAC3 polypeptide indicates that said mammal has a mutation in a FREAC3 gene, wherein the presence of said mutation is an indication that said mammal has an increased likelihood of developing a disease of the eye.

8. The method of claim 1, wherein said mammal is a human.

9. Nucleic acid encoding mutant FREAC3, wherein said nucleic acid has at least one mutation, wherein said mutation is an indication that a mammal from which said nucleic acid is derived has an increased likelihood of developing glaucoma.

10. A method of detecting a compound useful for the prevention or treatment of a disease of the eye, said method comprising assaying transcription levels of a reporter gene operably linked to a promoter, said promoter comprising a FREAC3 binding site, said method comprising the steps of:

- 15 (a) exposing said reporter gene to said compound, and
(b) assaying said reporter gene for an alteration in reporter gene activity relative to a reporter gene not exposed to said compound.

11. The method of claim 1, wherein said disease of the eye is glaucoma.

12. The method of claim 7, wherein said disease of the eye is glaucoma.

13. The method of claim 10, wherein said disease of the eye is glaucoma.

14. A method of treating a disease of the eye by *in vivo* gene therapy, said method comprising introducing into the cells of the eye a nucleic acid that
5 encodes wild-type FREAC3, wherein said nucleic acid is operably linked to regulatory sequences for expression of said FREAC3, wherein said regulatory sequences comprise a promoter, and wherein said expression of said FREAC3 is sufficient to ameliorate symptoms of said disease.

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ADD B3
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ADD E1

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